

APOB 3' VNTR POLYMORPHISM IN SOUTHERN AFRICAN POPULATIONS

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INTRODUCTION

Inter-ethnic differences in allele frequency distributions of VNTR containing loci exist and should be taken into account when using such systems for forensic purposes. The aim of the present study has been to determine whether significant differences in apoB 3' VNTR allele frequency distribution are detectable between southern African Negroids and Caucasoids and between subdivisions of these groups.



Fig. 1. A simplified representation of the approximate locations of the non-Caucasoid populations studied. The European Caucasoid and the Asian Indian populations were sampled in Johannesburg.

MATERIALS AND METHODS

The apoB 3' VNTR alleles of 653 individuals from 16 southern African populations were amplified by the polymerase chain reaction (Boerwinkle *et al* 1989) and sized by electrophoresis in agarose gels (Marques 1992). Each of the 16 population samples was checked for possible deviation from Hardy-Weinberg equilibrium using a bootstrapping method described by Devlin and Risch (1992a). The allele frequency distributions of the pooled Negroid and Caucasoid groups, subgroups and ethnic-groups were compared by calculating the Hellinger distance, H , (Devlin and Risch 1992b) between them.

$$H = \sqrt{\left(\sum_{i=1} \sqrt{\pi_i^{(1)}} - \sqrt{\pi_i^{(2)}} \right)^2}$$

where $\pi_i^{(1)}$ and $\pi_i^{(2)}$ are the frequencies of allele "i" in the two populations being compared. The significance of each Hellinger distance was then assessed by combining the data from the two samples being compared and creating two pseudo-samples (with the same number of alleles as the original samples) by selecting alleles at random from the pooled sample. The Hellinger distance between the two pseudo-samples was then calculated and compared to the true Hellinger distance. This process was repeated up to 500 times. If the simulated distance exceeded the true distance in fewer than 5% of trials, the difference between the actual population samples was judged to be significant.

RESULTS AND DISCUSSION

The genotype frequencies of all the ethnic-group samples were consistent with Hardy-Weinberg expectation (results not shown). The allele frequency distributions of some of the ethnic-groups sampled are shown in Figure 2.

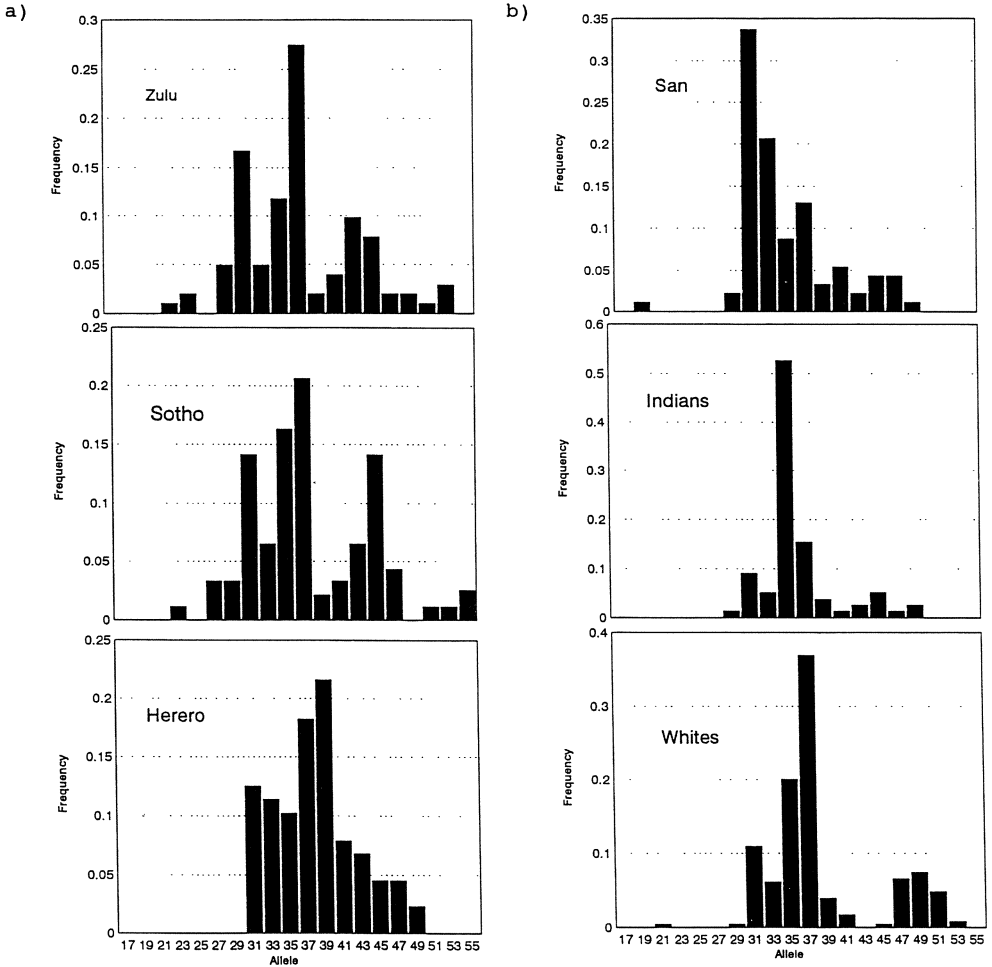


Fig. 2. Allele frequency distributions of representative ethnic groups: (a) Negroid groups. (b) Khoisan and Caucasoid groups.

The Hellinger distance between the pooled Negroid and Caucasoid samples is shown in Table 1a; the distance was found to be significant. The Caucasoid sample was divided into a European gentile, an Ashkenazi-Jewish and an Asian Indian subgroup and these subgroups compared to each other (Table 1b). The two main Caucasoid groups viz. the European gentiles and Asian Indians have significantly different allele frequency distributions. The Ashkenazi-Jewish sample did not differ significantly from the European gentile subgroup but did differ from the Indian group.

An attempt was then made to determine whether significant differences were apparent among the major Negroid subgroups. The following subgroups were constituted by pooling the data from various ethnic-groups on the basis of linguistic affiliation and geographic proximity (the ethnic groups of each subgroup are given in parenthesis):

1. Nguni (Zulu, Xhosa, Swazi and Tsonga)
2. Sotho-Tswana (Sotho, Tswana and Pedi)
3. Lemba-Venda (Lemba and Venda)
4. Herero-Dama (Herero and Dama)
5. Khoisan (Nama and San).

